

Contents



Original articles

- 241** Microarray based comparative genomic hybridisation (array-CGH) detects submicroscopic chromosomal deletions and duplications in patients with learning disability/mental retardation and dysmorphic features C Shaw-Smith, R Redon, L Rickman, M Rio, L Willatt, H Fiegler, H Firth, D Sanlaville, R Winter, L Colleaux, M Bobrow, N P Carter
- 249** Genomic imbalances in mental retardation M Kriek, S J White, M C Bouma, H G Dauwse, K B M Hansson, J V Nijhuis, B Bakker, G-J B van Ommen, J T den Dunnen, M H Breuning
- 256** Gene-gene interaction in folate-related genes and risk of neural tube defects in a UK population C L Relton, C S Wilding, M S Pearce, A J Laffling, P A Jonas, S A Lynch, E J Tawn, J Burn
- 261** Synergy between the C2 allele of transferrin and the C282Y allele of the haemochromatosis gene (*HFE*) as risk factors for developing Alzheimer's disease K J H Robson, D J Lehmann, V L C Wimhurst, K J Livesey, M Combrinck, A T Merryweather-Clarke, D R Warden, A D Smith

Short reports

- 266** A locus for spondyloraprotarsal synostosis syndrome at chromosome 3p14 C Steiner, N Ehtesham, K D Taylor, E Sebald, R Cantor, L M King, X Guo, T Hang, M S Hu, J-R Cui, B Friedman, D Norato, J Allanson, C Honeywell, G Mettler, F Field, R Lachman, D H Cohn, D Krakow
- 270** Absence of *BRAF* mutations in UV-protected mucosal melanomas R H Edwards, M R Ward, H Wu, C A Medina, M S Brose, P Volpe, S Nussen-Lee, H M Haupt, A M Martin, M Herlyn, S R Lessin, B L Weber
- 273** Homozygosity mapping of a third Joubert syndrome locus to 6q23 C Lagier-Tourenne, E Boltshauser, N Breivik, M Gribaa, C Bétard, C Barbot, M Koenig

Letters to JMG

- 278** Evaluation of widely used models for predicting *BRCA1* and *BRCA2* mutations F Marroni, P Aretini, E D'Andrea, M A Caligo, L Cortesi, A Viel, E Ricevuto, M Montagna, G Cipollini, S Ferrari, M Santarosa, R Bisegna, J E Bailey-Wilson, G Bevilacqua, G Parmigiani, S Presciuttini
- 286** No association between a previously reported *OLR1* 3' UTR polymorphism and Alzheimer's disease in a large family sample L Bertram, M Parkinson, K Mullin, R Menon, D Blacker, R E Tanzi
- 289** Detection of cell free placental DNA in maternal plasma: direct evidence from three cases of confined placental mosaicism H Masuzaki, K Miura, K-i Yoshiura, S Yoshimura, N Niikawa, T Ishimaru
- 293** Mitochondrial DNA haplogroups influence the Friedreich's ataxia phenotype M Giacchetti, A Monticelli, I De Biase, L Pianese, M Turano, A Filla, G De Michele, S Cocozza
- 296** Primary open angle glaucoma is associated with a specific *p53* gene haplotype T Ressiniotis, P G Griffiths, M Birch, S Keers, P F Chinnery
- 299** Isolated congenital anosmia locus maps to 18p11.23-q12.2 M Ghadami, S Morovati, K Majidzadeh-A, E Damavandi, G Nishimura, A Kinoshita, P Pasalar, K Komatsu, M T Najafi, N Niikawa, K Yoshiura
- 304** Novel lamin A/C gene (*LMNA*) mutations in atypical progeroid syndromes A B Csoka, H Cao, P J Sammak, D Constantinescu, G P Schatten, R A Hegele
- 309** Novel association of hypertrophic cardiomyopathy, sensorineural deafness, and a mutation in unconventional myosin VI (*MYO6*) S A Mohiddin, Z M Ahmed, A J Griffith, D Tripodi, T B Friedman, L Fananapazir, R J Morell
- 315** A novel locus for late onset amyotrophic lateral sclerosis/motor neurone disease variant at 20q13 A L Nishimura, M Mitne-Neto, H C A Silva, J R M Oliveira, M Vainzof, M Zatz

contd...

**NEW
ONLINE
SUBMISSION**

**GO TO
WEBSITE**

TO SUBMIT YOUR
MANUSCRIPT

...contd

Miscellanea

- 248** Echo
- 285** Echo
- 288** Echo
- 320** Correction

Electronic letters

- e38** *DBH* gene variants that cause low plasma dopamine β hydroxylase with or without a severe orthostatic syndrome J Deinum, G C H Steenbergen-Spanjers, M Jansen, F Boomsma, J W M Lenders, F J van Iltersum, N Hück, L P van den Heuvel, R A Wevers
- e39** Genetic and functional evaluation of an interleukin-12 polymorphism (*IDDM18*) in families with type 1 diabetes R Bergholdt, P Ghandil, J Johannesen, O P Kristiansen, I Kockum, H Luthman, K S Rønningen, J Nerup, C Julier, F Pociot
- e40** Assessment of association between variants and haplotypes of the remaining *TBX1* gene and manifestations of congenital heart defects in 22q11.2 deletion patients A Rauch, K Devriendt, A Koch, R Rauch, M Gewillig, C Kraus, M Weyand, H Singer, A Reis, M Hofbeck
- e41** Mitochondrial DNA haplogroup distribution within Leber hereditary optic neuropathy pedigrees P Y W Man, N Howell, D A Mackey, S Nørby, T Rosenberg, D M Turnbull, P F Chinnery
- e42** A frequent keratin 8 p.L227L polymorphism, but no point mutations in keratin 8 and 18 genes, in patients with various liver disorders M Hesse, T Berg, B Wiedenmann, U Spengler, R P Woitas, T M Magin
- e43** Intranuclear inclusions in neural cells with premutation alleles in fragile X associated tremor/ataxia syndrome F Tassone, R J Hagerman, D Garcia-Arocena, E W Khandjian, C M Greco, P J Hagerman
- e44** Comparison of motivations and concerns for genetic testing in hereditary colorectal and breast cancer syndromes J Balmaña, E M Stoffel, K M Emmons, J E Garber, S Syngal
- e45** Robust fragile X (CGG)_n genotype classification using a methylation specific triple PCR assay Y Zhou, H-Y Law, C D Boehm, C-S Yoon, GR Cutting, I S L Ng, S S Chong

- e46** FRG1P is localised in the nucleolus, Cajal bodies, and speckles S van Koningsbruggen, R W Dirks, A M Mommaas, J J Onderwater, G Deidda, G W Padberg, R R Frants, S M van der Maarel
- e47** Screening of the 1 Mb *SOX9* 5' control region by array CGH identifies a large deletion in a case of campomelic dysplasia with XY sex reversal R Pop, C Conz, K S Lindenberg, S Blesson, B Schmalenberger, S Briault, D Pfeifer, G Scherer

Online mutation reports

- e48** Automated comparative sequence analysis identifies mutations in 89% of NF1 patients and confirms a mutation cluster in exons 11–17 distinct from the GAP related domain C Mattocks, D Baralle, P Tarpey, C French-Constant, M Bobrow, J Whittaker
- e49** Functional analysis of novel *SLC11A1* (*NRAMP1*) promoter variants in susceptibility to HIV-1 H Donninger, T J Cashmore, T Scriba, D C Petersen, E Janse van Rensburg, V M Hayes
- e50** *FKRP* (826C>A) frequently causes limb-girdle muscular dystrophy in German patients M C Walter, J A Petersen, R Stucka, D Fischer, R Schröder, M Vorgerd, A Schroers, H Schreiber, C O Hanemann, U Knirsch, A Rosenbohm, A Huebner, N Barisic, R Horvath, S Komoly, P Reilich, W Müller-Felber, D Pongratz, J S Müller, E A Auerswald, H Lochmüller
- e51** Common origin of the Val30Met mutation responsible for the amyloidogenic transthyretin type of familial amyloidotic polyneuropathy H Ohmori, Y Ando, Y Makita, Y Onouchi, T Nakajima, M J M Saraiva, H Terazaki, O Suhr, G Sobue, M Nakamura, M Yamaizumi, M Munar-Ques, I Inoue, M Uchino, A Hata
- e52** Genomic organisation of the UDP-N-acetylglucosamine-1-phosphotransferase gamma subunit (GNPTAG) and its mutations in mucopolidosis III A Raas-Rothschild, R Bargal, O Goldman, E Ben-Asher, J E M Groener, A Toutain, E Stemmer, Z Ben-Neriah, H Flusser, F A Beemer, M Penttinen, T Olander, A J J T Rein, G Bach, M Zeigler